

Rabbit Anti-C2orf71 antibody

SL15158R

Product Name:	C2orf71
Chinese Name:	2号染色体开放阅读框71抗体
Alias:	Chromosome 2 open reading frame 71; Uncharacterized protein C2orf71; CB071 HUMAN.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Rat,Dog,Cow,Sheep,
Applications:	WB=1:500-2000ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800ICC=1:100-500IF=1:100-500 (Paraffin sections need antigen repair) not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	140kDa
Cellular localization:	The nucleus
Form:	Lyophilized or Liquid
Concentration:	lmg/ml
immunogen:	KLH conjugated synthetic peptide derived from human C2orf71:701-800/1288
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20 °C. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.
PubMed:	<u>PubMed</u>
Product Detail:	The protein encoded by this gene is highly expressed in photoreceptors and may associate with the primary cilium of the outer segment. The encoded protein appears to undergo post-translational lipid modification. Nonsense and missense variants of this gene appear to cause a recessive form of retinitis pigmentosa. [provided by RefSeq, Jun 2010]

Function:

May play an important role in the development of normalvision.

Subcellular Location:

Cell projection, cilium, photoreceptor outersegment.

Tissue Specificity:

Specifically expressed in retina.

DISEASE:

Defects in C2orf71 are the cause of retinitis pigmentosatype 54 (RP54) [MIM:613428]. A retinal dystrophy belonging to the group of pigmentary retinopathies. RP is characterized by retinalpigment deposits visible on fundus examination and primary loss ofrod photoreceptor cells followed by secondary loss of conephotoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually centralvision as well.

SWISS:

A6NGG8

Gene ID:

388939

Database links:

Entrez Gene: 388939 Human

Omim: 613425 Human

SwissProt: A6NGG8 Human

Important Note:

This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.